Questions and Answers About Living With Beckwith-Wiedemann Syndrome

1. What is Beckwith-Wiedemann Syndrome (BWS)?

Beckwith-Wiedemann Syndrome (BWS) is a rare overgrowth syndrome that occurs in approximately 1 in every 15,000 births. A small number of infants and children with BWS develop cancer.

Most children have only a few of the many distinct characteristics of the syndrome. The most common characteristics, which can range from mild to severe, are:

- Large tongue (macroglossia)
- Overall large body size with large organs
- Abdominal wall defects
- Above-average birth weight
- Uneven growth of limbs or organs (hemihypertrophy)
- Ear lobe creases or pits behind the upper ear
- Low blood sugar (hypoglycemia) shortly after birth
- Difficulty swallowing and eating
- Hearing loss
- Speech defects
- Occasional behavior abnormalities

About 10 percent of children with BWS develop cancer (or 10 out of 100 children with BWS). Studies suggest that the period of highest risk for developing cancer is before the age of 4 years. The most common types of cancer that occur in children with BWS are Wilms' tumor (kidney cancer) and hepatoblastoma (liver cancer). Other types of cancer, which occur more rarely, include adrenocortical carcinoma (cancer of the adrenal glands), neuroblastoma (cancer of the autonomic nervous system), and rhabdomyosarcoma (cancer of the muscle).

Children with BWS who have hemihypertrophy may have an increased risk of developing cancer. Children who have very large kidneys (almost double the size usually found at a given age) appear to be at greater risk for Wilms' tumor.

2. What kinds of care do children with BWS need?

Most children with BWS have the same needs as their brothers and sisters. Others have various special needs that change at different stages of growth and development.

Special Care for Newborns:

Newborns must have their blood glucose levels checked a few times after birth to make certain the levels are normal, and remain so, over the first few days. Newborns must also be evaluated for defects in the abdominal wall such as hernias (bulging of an organ or body part through the tissue that usually contains it). Sometimes such defects require surgery. Children who have large tongues may have eating and breathing problems. A speech therapist can offer valuable advice on feeding techniques. Hospital staff and social workers can provide support and resource information. A genetic counselor may also be helpful at this time.

Special Care in Infancy and Early Childhood:

Depending on their symptoms, infants and children with BWS may need special care. Members of the health care team may include the following:

- **Speech specialists**, who can check for possible difficulties with chewing, swallowing, choking, and breathing, in addition to speech problems.

- **Audiologists**, who can check for hearing loss.

- **Dietitians**, who can provide comprehensive nutrition services to children who are unable to eat properly.

- **Occupational and physical therapists**, who can help children with problems resulting from uneven growth of their arms or legs, or with gross motor skills such as crawling and walking. (Children with BWS may also need lifts in their shoes to make their legs equal in length.)

  Occupational and physical therapists can also treat sensory integration disorder, which can make a child unusually sensitive to touch and movement and trigger unusual behaviors.

- A **craniofacial team**, consisting of a plastic surgeon, an audiologist, a dentist, and/or an oral surgeon, should be consulted to evaluate any difficulties that may exist due to having a large tongue, and to decide if tongue reduction surgery would be beneficial.

Some children with BWS need help for behavior problems. It is not well established, however, whether their behavioral problems are actually part of BWS.

Cancer Screening in Infancy and Early Childhood:

Infants and small children with BWS need frequent and regular screening for cancer. Doctors generally recommend that BWS children have an abdominal ultrasound at least every 3 months until they are 7 or 8 years old to check for early stage Wilms' tumor. Ultrasound is a test that bounces sound waves off tissues and internal organs and changes the echoes into pictures (sonograms).

Blood levels of alpha fetoprotein (AFP) should be checked every 6 to 12 weeks until children are 3 or 4 years old. (An elevated level of AFP can suggest the presence of liver cancer.)

For screening to be effective at detecting cancers at an early stage, it is important to screen every 3 months. The risk of cancer decreases with time, particularly beyond the age of 8 years. Screening beyond that age is of uncertain value and is thus not routinely recommended. Nonetheless, for some parents, the reassurance of continued screening may warrant its use.

Checkups in Older Childhood:

The characteristics of BWS can vary over time. For instance, as children develop, their normal appearance may make it difficult to discern if their tongues are large. Differences between the length of each leg may increase or decrease with time. These changes require periodic evaluation and new lifts in the shoes. In general, however, children outgrow the visible signs of BWS by adolescence.

Although BWS is a lifelong condition, most children outgrow most of the characteristics of the syndrome by adulthood.

3. What is the best way to manage the care of children with BWS?

Children with BWS need regular check-ups and, depending on their symptoms, may need treatment in the areas listed above. Because BWS is a rare syndrome, parents must often educate others with whom their child comes into contact. Friends and relatives are likely to be more accepting and supportive if they understand the child's difficulties. Teachers may need the parents' guidance to understand the child's needs. Sometimes even doctors are not familiar with the syndrome. Also, health insurers may need to be informed so that they will cover the cost of needed services.

Occasionally, other people will be curious about the child's unusual features. Parents may worry about their child's reactions to questions or comments, but children with BWS usually handle the attention well.

4. What resources are available to help parents?

Some of the required evaluations and treatments are expensive. Health insurance companies generally cover most of the tests, but some insurers may deny coverage or pay for tests such as ultrasound less frequently than is recommended. In some cases, parents have appealed their insurers' decisions and been granted coverage.

Parents may also apply to their state and local health departments for services for children with special needs. Some states offer genetic services through their newborn screening or maternal and child health programs, which can be contacted through the state health department.

Children with BWS who are under 5 years old may benefit from participation in early intervention programs that are sponsored by the local public schools. (The age varies depending on the school district.) Some children will need special services throughout their education. Parents may find it helpful to talk to other parents, and professionals, who are familiar with the services available in the local school district.

Financial assistance for medical expenses may be available in the form of Medicaid, the Supplemental Security Income Program (SSI), health care services offered under the Children with Special Health Care Needs (CSHCN) provisions of the Social Security Act, and/or the State Children's Health Insurance Program (SCHIP).

- Medicaid, a jointly funded, Federal-State health insurance program for people who need financial assistance for medical expenses, is coordinated by the Health Care Financing Administration (HCFA). Information about coverage is available from local state welfare offices, state public health departments, state social services agencies, or the state Medicaid office. More information can be found on the HCFA Web site at http://www.hcfa.gov/medicaid/medicaid.htm on the Internet.

- Supplemental Security Income (SSI) is a Federal program administered by the Social Security Administration (SSA). Strict financial criteria must be met, and each application is decided individually. A child is considered impaired or disabled if there is a physical or mental condition or conditions that can be medically proven and that result in "marked and severe functional limitations." The condition or conditions must be expected to last for at least 1 year or result in the child's death.

  To be eligible, a child's impairment or disability must fit into a specific category in SSI's Listing of Impairments, or it must be determined to be medically or

functionally equivalent to an impairment on the list. To determine if the child's impairment causes "marked and severe functional limitations," the SSI disability evaluation team obtains information from people who are familiar with the child's condition and how it affects the child's ability to function on a day-to-day basis and over time. The sources include doctors and other health professionals who treat the child, teachers, counselors, therapists, and social workers. Once deemed eligible for SSI, a child's continued participation in the program is reviewed on a regular basis.

Parents can contact their local SSA office for more information about eligibility. SSA offices are listed in the Government section of the white pages. The SSA Web site also has information about the SSI program. It can be accessed at http://www.ssa.gov on the Internet.

- Children who are eligible for SSI are referred for health care services under the Children with Special Health Care Needs (CSHCN) provisions of the Social Security Act. CSHCN programs are usually administered through state health agencies. The Federal Maternal and Child Health Bureau, which is part of the Health Resources and Services Administration, oversees the states' CSHCN programs.

  There are differences from state to state, but most CSHCN programs provide specialized services through arrangements with clinics, private offices, hospitals, or community agencies. CSHCN programs may be called Children's Special Health Services, Children's Medical Services, Handicapped Children's Program, or another name, depending on the state.

  Even if a child is <u>not</u> eligible for SSI, a CSHCN program may be able to help. Local health departments or social services offices can provide information about a state's CSHCN program.

- The State Children's Health Insurance Program (SCHIP) is a Federal-State partnership that offers low-cost or free health insurance coverage to children in families with limited income. More information about SCHIP can be obtained by calling the toll-free Insure Kids Now Hotline at 1–877–KIDS–NOW (1–877–543–7669). The Insure Kids Now Web site is located at http://www.insurekidsnow.gov on the Internet.

Parents whose children have been denied SSI or special school services may find it helpful to seek legal advice. Low-income families can obtain advice from local Legal Services offices. If an appeal for SSI is successful, payments will be made retroactively.

Programs such as the NCI's Cancer Information Service (CIS) (see below) can provide information if a child is diagnosed with cancer.

5. How can parents of children with BWS reach out to other parents?

Many parents share their experiences with BWS with trusted family and friends. If other BWS families live in the community, they can form a support group. Support groups provide opportunities for parents to share information and increase awareness of the syndrome. Groups also offer an environment of acceptance and can lessen feelings of isolation.

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Sources of National Cancer Institute Information

Cancer Information Service

Toll-free: 1–800–4–CANCER (1–800–422–6237)
TTY (for deaf and hard of hearing callers): 1–800–332–8615

NCI Online

Internet

Use http://cancer.gov to reach NCI's Web site.

CancerMail Service

To obtain a contents list, send e-mail to cancermail@icicc.nci.nih.gov with the word "help" in the body of the message.

CancerFax® fax on demand service

Dial 301–402–5874 and listen to recorded instructions.

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